Seminars in Fetal & Neonatal Medicine 19 (2014) 306–311

Contents lists available at ScienceDirect

Seminars in Fetal & Neonatal Medicine

journal homepage: www.elsevier.com/locate/siny

Review

Ethical language and decision-making for prenatally diagnosed lethal malformations

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Keywords:
Infant
Newborn
Fatal outcome
Ethics
Medical futility
Trisomy
Fetal termination

SUMMARY

In clinical practice, and in the medical literature, severe congenital malformations such as trisomy 18, anencephaly, and renal agenesis are frequently referred to as 'lethal' or as 'incompatible with life'. However, there is no agreement about a definition of lethal malformations, nor which conditions should be included in this category. Review of outcomes for malformations commonly designated 'lethal' reveals that prolonged survival is possible, even if rare. This article analyses the concept of lethal malformations and compares it to the problematic concept of 'futility'. We recommend avoiding the term 'lethal' and suggest that counseling should focus on salient prognostic features instead. For conditions with a high chance of early death or profound impairment in survivors despite treatment, perinatal and neonatal palliative care would be ethical. However, active obstetric and neonatal management, if desired, may also sometimes be appropriate.

1. Introduction

Antenatal screening, particularly the use of routine mid-trimester ultrasound screening, has altered the diagnosis of major congenital malformations. As a result, in many parts of the world it is now uncommon for major malformations to be discovered at birth [1]. Antenatal diagnosis potentially allows targeted diagnostic testing, planning of delivery, counseling and education of couples, and earlier postnatal intervention for newborns with congenital malformations [2]. However, antenatal diagnosis may identify severe abnormalities where treatment is unavailable, or unlikely to be successful, and where fetal or neonatal death is a likely outcome. Such cases are often referred to as 'lethal malformation' (LM) (Box 1).

The diagnosis of LM is often said to carry ethical and legal implications for management during pregnancy, delivery, and postnatally [3–6]. For example, it may permit obstetric management focused on maternal well-being rather than on fetal survival, termination of pregnancy (including late in pregnancy), or non-resuscitation at birth [7]. But what do we mean when we refer to a malformation as 'lethal'? Which conditions fit into this category? What are the ethical implications of diagnosis of LM?

2. The concept of 'lethal malformation'

The word 'lethal' is derived from the Latin 'letalis' (deadly), and related to a Greek word meaning 'oblivion,' referring to the myth that the souls of the dead forgot their lives on Earth after drinking the waters of the River Lethe. Conventionally, 'lethal' is used to describe something (e.g. an action or agent) that will cause death [8].

In theory, there are several different ways to interpret the description of a malformation as lethal (Box 2).

A review of the published literature on LM revealed no consensus on which of these definitions should be applied [9]. The first definition does not apply to any of the commonly cited LMs, and is not one found in the literature. The second definition is probably the most plausible and the most frequently encountered [5,10–13]. Chervenak and McCullough endorse this definition: 'a lethal condition, properly understood, invariably leads to death, i.e., there is no effective treatment that will prevent a condition, disease, or injury from causing death in the near future' [14]. However, this definition does not apply to any of the malformations that are often described as lethal. Some papers have used the third definition [15–18]. This raises a question about how high a chance of

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http://dx.doi.org/10.1016/j.siny.2014.08.007

Box 1
Malformations most frequently described as 'lethal' conditions [9].

Potter's syndrome/renal agenesis
Anencephaly/acrania
Thanatophoric dwarfism
Trisomy 13 or 18
Holoprosencephaly

death is sufficient to fit into a lethal category. The cited proportion ranges from 50% to 'almost all' [16,19]. There is neither agreement about the correct proportion, nor any obvious way to determine where the cut-off should lie. The fourth definition is used in some epidemiologic studies of neonatal mortality [20–22]. However, it appears far too broad to correspond to the way that LM is used by obstetricians and neonatologists.

3. Which malformations are lethal?

Although Box 1 lists the most frequently cited LMs, more than 25 conditions are included in different lists [9]. No condition was present on all lists, and there was considerable variation.

What is the outcome for these malformations? Table 1 presents an attempt to estimate outcome; however, the values cited are necessarily imprecise. High proportions of affected pregnancies are terminated [47]. Since these conditions are associated with high fetal death rate, survival rate also varies with the gestational age at the time of diagnosis. Postnatal survival is also difficult to estimate because of selection bias in published cohorts, and because of the problem of self-fulfilling prophecies [48,49]. Where a large proportion of infants receive palliative care after birth, a high mortality rate is inevitable [9].

What is clear from Table 1 is that survival of at least six months has been described in all of the conditions frequently cited as lethal. Most strikingly, this includes both anencephaly and bilateral renal agenesis. There has been a very recently published case report of an infant in the USA with Potter syndrome who was treated with antenatal amnio-infusion and neonatal renal dialysis and who survived to be listed for renal transplantation at one year of age.

Box 2
Possible definitions of a 'lethal congenital malformation'.

1. Fetal death: a condition that invariably leads to death in-utero
2. Fetal death/neonatal death: a condition that invariably leads to death either in utero or in the newborn period regardless of treatment
3. Usual fetal/neonatal death: a condition that leads to death in utero or in the newborn period in most cases
4. Associated with death: a condition that leads to fetal or neonatal death in some cases

4. The significance of a 'lethal diagnosis': the examples of trisomy 18 and 13

The severe autosomal trisomies, 18 (Edwards syndrome; T18) and 13 (Patau syndrome; T13), are frequently described as lethal [18,50–53]. Yet, recent population cohort studies show that more than half of affected live-born infants survive for more than a week, and up to 20% survive for more than a year [18,29]. In a large US series including 52,262 very low birth weight infants, 11% of infants with T13 and 9% of infants with T18 survived to discharge [54]. It is possible that even these values represent an underestimate of potential survival rates, since in parts of the world where cardiac surgery is offered to infants with T13 or T18, one-year survival rates as high as 50% have been reported [55].

Why does it matter if these conditions are described as lethal? The first reason to be concerned about this terminology is its potential for misunderstanding and miscommunication. We surveyed more than 1000 obstetricians from the UK, Australia, and New Zealand about the perinatal management of T18 [56]. The overwhelming majority (85%) of obstetricians regarded T18 as a lethal malformation. More than 50% regarded T18 as 'incompatible with life'. We did not ask obstetricians whether they would use these terms in counseling, but a survey of parents from T13/T18 support groups found that 93% had been told by health professionals that their child's condition was 'lethal or incompatible with life' [57]. This contrasts with the evidence summarized above, and with obstetricians' own understanding about survival. Three-quarters of respondents estimated that at least 5% of affected infants would survive for more than one year if treatment were provided [56].

Qualitative studies and narratives from parents of infants with T13 or T18 describe feelings of anger and disillusionment and a sense of being misled by health professionals as well as by the language used [58–61]. Many parents reported that health care providers were unable to look beyond adverse statistics [57]. Furthermore, the Internet has provided families with the ability to do their own research and encounter alternate perspectives on their child's condition. Within seconds of searching for 'trisomy 18' a parent may see pictures of many older children with trisomy 18, smiling and happy, strong evidence against 'incompatibility with life'. If they have been told by their doctor that trisomy 18 is always lethal, there may be repercussions for the family's ongoing capacity to trust health professionals [44].

Another reason to be concerned about denoting a condition such as T18 as 'lethal' is because of a worry that this language contains concealed value judgments about the quality of life of surviving infants [31,49]. Eighty percent of obstetricians in our survey believed that T18 was not compatible with a 'meaningful life' [56]. Labeling a condition as 'lethal' may also risk taking decision-making from the parents [31,49]. In our survey, 23% of obstetricians would never discuss or offer fetal monitoring in labour for women after an antenatal diagnosis of T18, and 28% would never offer caesarean section for fetal distress [56]. In the parent-support group study, two-thirds of parents reported feeling pressure to terminate their pregnancy [57].

There is a concept in medical ethics that shares some features with that of LM: the concept of 'medical futility' [9,19]. Medical futility emerged in the 1990s as a potential way to resolve disputes between patients and doctors about life-sustaining treatment [62]. It reflected a perceived need by medical professionals to limit patient autonomy and to justify a decision not to provide treatment that had been requested [62]. The basic idea was that although it was important to respect patients' views about treatment, health professionals were not obligated to provide futile treatment [63]. However, the concept of futility has fallen out of favour because of a